
Targeted resequencing of 52 bone marrow failure genes in patients with aplastic anemia reveals an increased frequency of novel variants of unknown significance only in SLX4

Laura C. Collopy,¹ Amanda J. Walne,¹ Tom J. Vulliamy,^{1*} and Inderjeet S. Doka^{1,2†}

¹Centre for Paediatrics, Blizard Institute, Barts and The London School of Medicine and Dentistry, Queen Mary University of London, London; and ²Barts Health NHS Trust, London, UK.

Correspondence: t.vulliamy@qmul.ac.uk.

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Supplementary Table – 11 BMF syndromes and their associated genes included in the study

BMF syndrome	Gene associated	Chromosomal coordinates	BMF syndrome	Gene associated	Chromosomal coordinates		
Fanconi anemia (FA)	<i>BRCA2</i>	chr13:32,945,093-32,953,652	Diamond-Blackfan anemia (DBA)	<i>RPL5</i>	chr1:93,301,784-93,306,565		
	<i>BRIP1</i>	chr17:59,762,133-59,858,366		<i>RPL11</i>	chr1:24,018,269-24,022,915		
	<i>FANCA</i>	chr16:89,806,149-89,831,474		<i>RPL26</i>	chr17:8,280,834-8,286,565		
	<i>FANCB</i>	chrX:14,861,529-14,891,184		<i>RPL35A</i>	chr3:197,677,095-197,682,721		
	<i>FANCC</i>	chr9:97,861,336-98,079,536		<i>RPS7</i>	chr2:3,625,381-3,628,509		
	<i>FANCD2</i>	chr3:10,091,058-10,143,614		<i>RPS10</i>	chr6:34,385,231-34,393,902		
	<i>FANCE</i>	chr6:35,420,138-35,434,881		<i>RPS17</i>	chr15:82,821,161-82,906,047		
	<i>FANCF</i>	chr11:22,644,079-22,647,387		<i>RPS19</i>	chr19:42,363,988-42,375,484		
	<i>FANCG</i>	chr9:35,075,462-35,079,898		<i>RPS24</i>	chr10:79,793,518-79,800,473		
	<i>FANCI</i>	chr15:89,787,194-89,860,362		<i>RPS26</i>	chr12:56,435,686-56,438,007		
	<i>FANCL</i>	chr2:58,387,243-58,468,515		Severe congenital neutropenia (SCN)	<i>CSF3R</i>	chr1:36,931,644-36,948,915	
	<i>FANCM</i>	chr14:45,644,274-45,670,093			<i>ELANE</i>	chr19:852,291-856,246	
	<i>PALB2</i>	chr16:23,614,483-23,652,678			<i>G6PC3</i>	chr17:42,148,098-42,153,712	
	<i>RAD51C</i>	chr17:56,769,963-56,772,241			<i>GFII</i>	chr1:92,940,318-92,952,433	
<i>SLX4</i>	chr16:3,650,622-3,661,585	<i>HAX1</i>	chr1:154,245,039-154,248,355				
Dyskeratosis congenita (DC)	<i>CTCI</i>	chr17:8,128,139-8,151,413	Congenital dyserythropoietic anemia (CDA)	<i>CDANI</i>	chr15:43,024,531-43,027,572		
	<i>DKC1</i>	chrX:153,991,031-154,005,964		<i>KLF1</i>	chr19:12,995,237-12,998,017		
	<i>NHP2</i>	chr5:177,576,465-177,580,961		<i>SEC23B</i>	chr20:18,488,188-18,542,059		
	<i>NOP10</i>	chr15:34,633,917-34,635,362	Wiskot Aldrich syndrome (WAS)	<i>WAS</i>	chrX:48,542,186-48,549,817		
	<i>RTEL1</i>	chr20:62,289,667-62,328,544		Barth syndrome	<i>TAZ</i>	chrX:153,640,181-153,650,063	
	<i>TERC</i>	chr3:169,482,398-169,482,848			WHIM syndrome	<i>CXCR4</i>	chr2:136,871,919-136,875,307
	<i>TERT</i>	chr5:1,278,756-1,295,162				Congenital amegakaryocytic thrombocytopenia (CAMT)	<i>MPL</i>
	<i>TINF2</i>	chr14:24,708,851-24,711,880					
	<i>USB1</i>	chr16:58,035,277-58,055,527					
	<i>WRAP53</i>	chr17:7,604,059-7,605,807					
Familial myelodysplasia leukemia (FML)	<i>AML1</i>	chr21:36,160,098-36,421,595					
	<i>CEBPA</i>	chr19:33,790,840-33,793,430					
	<i>GATA2</i>	chr3:128,198,265-128,212,030					
	<i>SRP72</i>	chr4:57,333,762-57,369,847					
Shwachman-Diamond syndrome (SDS)	<i>SBDS</i>	chr7:66,452,690-66,460,588					